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Risk Perception After CF Carrier Testing and Impact of the Test Result on Reproductive Decision Making

Lieve Denayer, Myriam Welkenhuysen, Gerry Evers-Kiebooms,* Jean-Jacques Cassiman, and Herman Van den Berghe
Center for Human Genetics, Leuven, Belgium

A correct interpretation of the result of a CF carrier test and a correct understanding of the risk of having a CF child is complicated by the limited sensitivity of the DNA test. The present paper addresses this problem, with special attention for the residual risk after a negative test result. Secondly, the impact of the test results on child bearing decisions is evaluated. Seventy percent of a sample of 200 subjects tested in the Center for Human Genetics in Leuven answered a mailed questionnaire and participated in an interview. Knowledge about the recessive transmission of CF was weak. In the case of both partners receiving a negative test result, more than half the subjects were not aware of the restrictions of DNA testing, and were convinced that there was no residual risk. Two thirds of the subjects from the 'carrier + negative test result couples' were aware of the residual risk of having a CF child. This risk did not hamper further reproduction. Carriers who were single or whose partner was not tested, had much difficulties to understand the risk. Although most of them had no idea about their exact risk level, it influenced their childbearing decisions to some extent. It was also shown that the own risk of having a CF child had an impact on attitudes toward reproduction in hypothetical situations with different risk levels. Am. J. Med. Genet. 69:422–428, 1997. © 1997 Wiley-Liss, Inc.

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INTRODUCTION

Cystic Fibrosis (CF) is the most common, potentially lethal autosomal recessive disorder in populations of Caucasian descent, with a carrier prevalence of one in 25 and a prevalence of affected persons of one in 2,500 live births. Since the identification of the CF gene carrier detection has been widely used within affected families. As most CF patients are born in families with no history of CF, population screening is the only way to reach most of the couples at risk of having a CF child. However, experiences with genetic testing have shown serious problems such as confusion, stigmatization, and discrimination when no comprehensive infrastructure provides education, informed consent, and counseling [Wilfond and Fost, 1992]. For CF, the situation is even more complicated because of the wide spectrum of mutations, the limited sensitivity of current tests and the resulting ambiguity of a negative test result [Tsui, 1994]. The potential anxiety caused in couples in which one partner is a carrier, while the other has a negative test result but cannot be excluded as a carrier of a rare CF mutation, is a major concern. Pilot programs for screening were initiated in several countries [Mennie et al., 1992; Watson et al., 1992; Kaplan et al., 1994]. However, in many other countries (including Belgium) the general policy has been adopted not to offer CF carrier testing actively to large groups. In the Center for Human Genetics in Leuven carrier testing has been requested most frequently by people with a family history of CF. The reactions of this group toward genetic testing should be studied before generalized population screening is considered.

For the group tested in Leuven, the overall effect of the test result on carriers' and non-carriers' feelings about themselves and on the feelings they attribute to most carriers and to most noncarriers has already been discussed in Evers-Kiebooms et al. [1994]. Additional psychometric testing at least one year after they received the test result showed no negative effect on CF carriers' self-image nor a lasting increase in anxiety [Denayer et al., 1996].

The present paper focusses on risk perception, with particular attention paid to the awareness of the residual risk in case of a negative test result. Secondly, the effect of knowing one's risk status on reproductive decision making is evaluated.

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*Correspondence to: Prof. Dr. Gerry Evers-Kiebooms, Center for Human Genetics, Herestraat 49 B-3000 Leuven, Belgium.

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MATERIALS AND METHODS

A mailed questionnaire was sent to all adult individuals (N = 200) who were tested in the Center for Human Genetics, Leuven between 1986 and 1992. During this period, both linkage analysis and direct testing had been used for CF carrier testing. Direct testing for the eight most frequent mutations (ΔF508, ΔI507, G542X, 1717-IG → A, R553X, G551D, W1282X and N1303K) detects about 85% of all CF mutations in the Belgian population [Cuppens et al., 1993]. Seventy percent of the tested individuals returned the questionnaire; 126 of them had a follow-up interview at least one month later. The questionnaire covered various aspects of carrier testing, and contained both open and multiple choice questions. We refer to Evers-Kiebooms et al. [1994] for more details about this extensive questionnaire.

The present paper reports on the sections concerning the understanding of recessive inheritance and the perception of the own risk of having a CF child after carrier testing, taking into consideration the result of the partner. Knowing that very few people can reproduce exact risk figures, a multiple choice question with five risk intervals was used. Since the categories had to be the same for all questions about risk, they had to be very broad (cf. Appendix). Immediately after answering the multiple choice question, the exact numerical figure had to be given. The questionnaire also investigated hypothetical decision making, should both partners have been told before marriage that they were carriers of the CF gene.

In a second stage of the study, all respondents were contacted again for an interview about the psychological implications of the test result and for testing the respondents' anxiety level and self concept [Denayer et al., 1996]. In the present paper, the test scores are used as correlates. Actual reproductive decisions were assessed by comparing the number of children they had at the moment of carrier testing with the number of children in the family at the time of the study. During the interview we also tried to determine the risk level that would influence the respondents' childbearing decisions, starting from their own post test risk level. To reach some uniformity in the answers, the respondents were asked to imagine that they had no children, but would like to have children, and that prenatal diagnosis was not available. If their actual risk level would make them hesitate about getting pregnant, gradually lower risk levels were explored, until reaching a risk level that would not have any influence; if their actual risk level would not make them hesitate, higher risk levels were considered until reaching the point where the risk would have an influence (hypothetical risk levels used: 1/100.000, 1/10.000, 1/1.000, 1/100, 1/10, 1/1). At the end of the interview, the respondent's correct risk status was always repeated. If needed, additional information was given, to guarantee an accurate understanding of test results.

For the analyses in the present paper eight individuals had to be excluded because of too many missing data. Table I gives a description of the sociodemographic characteristics of the sample. Males and females were equally represented. The median age was 29 years.

Two thirds of the subjects had a CF patient in their own family (19% sibs versus 45% more distant relatives), while 27% had a CF patient in their partner's family. Twelve persons (9%) were not related to a CF patient. The sample consisted of 42 CF carriers and of 88 individuals with a negative test result; one respondent had an inconclusive result. The carriers were all related to a CF patient. Eighty percent of the subjects had carrier testing through direct mutation testing; for 6% this was combined with a linkage study. Twenty percent of the subjects only had the results of a linkage study (these analyses were done before the discovery of the CF gene in 1989). Seventy-six of the 131 respondents in this study belonged to a couple of which both partners participated in the study. From the other 55 individuals, 12 were single at the time of the study; in 9 cases the partner was tested, but did not participate in this study; in 34 cases the partner was not tested. At the time of DNA-analysis, one to six years before, 43% were married but had no children; 28% were married and had children (more than half of them had only one child). To evaluate the influence of CF carrier testing on actual childbearing decisions, 'procreational units' served as the unit for observation. Therefore, one partner was discarded at random from those couples of which both partners participated in the study and the analyses were restricted to those subjects who wanted children at the time of carrier testing.

RESULTS

1. Risk Perception in a Population Tested for CF Carriership

1.1. Understanding of autosomal recessive transmission. The high risk of having a CF child when both parents are carriers of the CF gene was the best known item in this context. Seventy two percent of the respondents estimated it to be higher than 1 in 10. Fifty-one percent of the respondents knew the exact risk of having a CF child (1 in 4). The fact that there is no risk of having a CF child when neither parent is a CF carrier, was known by two thirds of the respondents (66%). Forty-nine percent were aware of the zero risk when only one parent is a CF carrier. Only 14% answered all three questions correctly. The a priori risk that a ran-

TABLE I. Sociodemographic Description of the Sample at the Time of the Study (N = 131)

Age	18–19 years	5%
	20–29 years	55%
	30–39 years	34%
	40 years and older	6%
Educational level	< High school	19%
	High school	33%
	> High school	36%
	University degree	12%
Marital status	Unmarried—no partner	9%
	Unmarried—partner	11%
	Married—no children	23%
	Married—one or more children	57%

dom person (someone who has no CF patient in his or her family) is a carrier of the CF gene was poorly known. One third (38%) had no idea about this risk. Another third (36%) severely underestimated the risk (<1 in 100). Only 12% knew that a random person has a risk between 1 in 20 and 1 in 30 of being a CF carrier.

Most of the respondents (63%) reported that the mechanism of autosomal recessive transmission was explained by a family member, while the others received the information from a medical doctor or did not remember having been informed about autosomal recessive transmission. The latter was the case for 7%.

1.2. Perception of the risk of having a CF child after carrier testing. Eighty-four of the subjects reported that they received the test result directly from a medical doctor, while 16% were informed indirectly through family members. Unfortunately we have no details about the exact information they received.

Most respondents (95%) correctly remembered whether they received a positive or negative test result. However, two carriers were convinced they received a negative result and 5 individuals with a negative test result could not remember the outcome of the test; however they remembered that they had been reassured about their risk of having a CF child. Only half (55%) of the entire sample remembered that they had been informed explicitly about the couple's risk of having a CF child.

For the analysis of the subjects' understanding of the post test risk, the sample was divided into four subgroups, according to the tested individual's objective risk of having a CF child, taking into consideration the (availability of the) partner's test result. Table II gives an overview of the risk as perceived by each subgroup.

1.2.1. Both partners tested negatively or the respondent tested negatively while nothing was known about the partner (N = 69): for the sake of simplicity these subjects will be referred to as 'NC × NC'. Their post test risk of having a CF child is smaller than the population risk. Less than one quarter (22%) indicated the correct answer category ('less than 1 in 1000') for their post test risk. A majority (58%) thought they had no risk at all. A significant correlation was found with 'trait anxiety' as measured by the Spielberger State Trait Anxiety Inventory (STAI): the higher trait anxiety, the higher the quantitative estimation (tau = .37, $p < .001$) and the qualitative evaluation (tau = .33, $p < .01$) of the residual risk.

TABLE II. Risk of Having a CF-Child After Carrier Testing

	NC × NC (N = 69)	C × NC (N = 35)	C × ? (N = 25)
Estimation of the risk[a]			
Don't know	17%	8%	20%
(1) More than 1/10	—	—	8%
(2) 1/10 to 1/100	—	—	**12%**
(3) 1/100 to 1/1.000	3%	**46%**	24%
(4) Less than 1/1.000	**22%**	20%	12%
(5) Zero	58%	26%	24%
Subjective evaluation			
No answer	4%	3%	4%
(1) Very high risk	—	—	—
(2) High risk	—	6%	24%
(3) Low risk	7%	28%	24%
(4) Very low risk	35%	57%	20%
(5) No risk at all	54%	6%	28%

[a]The correct category is bolded for each of the three groups. The exact numerical figure was not identical for each individual in the same group, because the number of mutations analysed was higher at the end of the study period.

1.2.2.One partner tested positive while the other tested negative: This subgroup ('C × NC') consists of 35 individuals. Their risk of having a CF child is higher than the priori risk in the population. In the 'C × NC' group, 27 subjects (77%) remembered having been informed about their risk as a couple. About half (16 subjects) situated their post test risk in the correct category: between 1 in 100 and 1 in 1000. A majority (57%) subjectively evaluated this risk to be 'a very low risk'.

1.2.3. One subject is a carrier of the CF gene, while nothing Is known about the partner: This third subgroup ('C × ?') consisted of 25 CF carriers who were single or whose partner was not tested. They had a risk of about 1 in 100 when they (their partners) would become pregnant without further testing. In the 'C × ?' group only one person remembered having been informed about his post test risk of having a CF child. This group gave very divergent answers, and the highest proportion of 'don't know' answers. In this group also Trait Anxiety as measured with the STAI correlated in a significant way with both the quantitative estimation (tau = .38, $p < .05$) and qualitative evaluation (tau = .37, $p < .05$) of the residual risk: the higher the score for trait anxiety, the higher one estimated and/or evaluated the residual risk to be.

1.2.4. Both partners are carrying the CF gene: This fourth subgroup ('C × C') corresponds to the highest risk level. Our study contained one couple in this situation. Both subjects remembered having been informed about their risk of having a CF child; they were perfectly aware of their 1 in 4 risk and evaluated it as a very high risk.

1.2.5. A comparison among groups: The Jonckheere test for ordered alternatives [Siegel & Castellan, 1988] confirmed the influence of the objective post test risk level, both on the quantitative estimation ($J^* = 3.48$, $P < .001$) and on the qualitative evaluation ($J^* = 3.82$, $P < .001$) of the risk: the lower the objective post test risk, the lower the subjective quantitative estimation and the subjective qualitative evaluation of the risk of having a CF child. Multiple comparisons (in which the 'C × C' group was omitted because of the small number) showed a significant difference in both the mean quantitative estimation and the mean qualitative evaluation of the risk between the first subgroup on the one hand and the second and third subgroup on the other hand: the post test risk of partners with a negative test result was experienced as lower than the post test risk of a 'carrier + negative test result couple' and than that of a carrier whose partner was not tested. The difference between both the mean quantitative estimation and the mean qualitative evaluation of the risk of the second and the third subgroup was not significant.

1.3. Awareness of the limited sensitivity of DNA testing. After a renewed explanation of the post test

risk by the interviewer, it was always checked whether the respondents had been aware of the limited sensitivity of the DNA test for CF carriership, implying that, after a negative test result, nobody can be completely sure not to be a CF carrier. This was the case for 43% of the 105 subjects who were tested through direct mutation analysis. However, the percentages were very different according to the risk group. In the 'NC × NC' group (N = 57) 37% was aware of the residual risk. In the 'C × NC' group (N = 33), for whom this question is most pertinent, 64% was aware of the residual risk while in the 'C × ?' group (N = 13) the percentage dropped to 23%.

A Kruskall-Wallis test showed significant differences between groups ($P < .01$). Multiple comparisons showed that significance was due to the 'C × NC' group, who was more often aware of the limitations of DNA testing than both other groups.

2. Reproductive Decision Making After Carrier Testing

The 'NC × NC' group consisted of 42 'procreational units'; 32 had one or more pregnancies after carrier testing; one couple wanted children but had fertility problems; nine others wanted children in a more remote future. The 'C × NC' group consisted of 16 'procreational units'; 12 had one or more children after carrier testing; four wanted children in a more remote future.

The 'C × ?' group consisted of 17 'procreational units'. For 4 of them, the procreational phase was over at the time of the study: two of these carriers had no children, the other two had one child. Three carriers planned a pregnancy in the future; only one of them intended to urge his partner to have a carrier test beforehand. Ten subjects had not reached the procreational phase; all but one intended to have their partner tested at the appropriate moment.

The 'C × C' couple had one pregnancy before direct mutation testing was possible (knowing one of them was a carrier for sure and the other, being a remote relative of a CF patient, had an increased risk of being a carrier), and a pregnancy with prenatal diagnosis later on. Both pregnancies resulted in an unaffected child. Both partners felt that the second pregnancy was much more difficult on the psychological level, and they decided 'not to tempt fate any more'. During the interviews the first two groups ('NC × NC' and 'C × NC') clearly had no problems with their childbearing decisions. They all perceived the risk of having a CF child after carrier testing as acceptable. In the last two groups ('C × ?' and 'C × C') however, the risk of having a CF child had played a more substantial part in reproductive decision making. Especially for carriers (detected by linkage analysis) who made childbearing decisions before testing their partner was possible, the decision had been very difficult. One carrier deliberately decided against a second child because of the risk, and felt very unhappy about this decision afterwards. A young carrier had been implored by his (meanwhile deceased) CF brother never to have children and felt obliged to follow this advice. He did not know that carrier testing had become a reality also for his partner. However, the two carriers who remained childless invoked other reasons than their risk of having a CF child. A substantial part of the subjects in the 'C × ?' group wanted children anyhow. One carrier was very easygoing about his risk, sure that it would not happen to him. Another one preferred not to know the carrier status of his partner, and to take the risk of having a CF child; his partner agreed with this decision. A young man who was still single explicitly said he hoped that a future partner would agree to 'take the risk' without knowing whether she was a CF carrier or not.

To investigate the influence of the post test risk on child bearing decisions in a more objective way, an analysis of covariance was performed on the couples having procreative intentions at the time of carrier testing. Because of the small number of observations, the 'C × C' group had to be omitted from this analysis. The dependent variable of this analysis was the number of children born after carrier testing. The amount of time elapsed since marriage and the number of children already born before carrier testing were controlled as covariates. The three groups did not differ significantly with regard to the number of children born after carrier testing, when the above-mentioned variables were controlled.

3. Attitudes Toward Reproduction in Hypothetical Risk Situations

3.1. Reproductive decision making in the hypothetical situation that both partners would have been informed that they were carriers of the CF gene. All respondents were asked how they would have reacted, had they been told before marriage that they had a 1 in 4 chance of having a CF child in every pregnancy. When asked if this would have been an obstacle for a marriage, 65% of the respondents told that this would surely not have been an impediment; 32% answered that this would probably not have been an obstacle. The others did not answer the question. Next, they were asked about the hypothetical impact on decision making about having children. Table III gives hypothetical decisions regarding having children, prenatal diagnosis and pregnancy termination for the group as a whole and for the three subgroups separately. A Kruskall-Wallis one-way analysis of variance showed a significant influence of the post test risk on hypothetical decisions about having children in a high risk situation. Multiple comparisons showed that the 'C × ?' group was responsible for this difference: they primarily opted for having own children, with or without prenatal diagnosis, while the 'NC × NC' group and the 'C × NC' group considered more easily remaining childless.

3.2. Evaluation of several risk levels. Figure 1 shows cumulative percents of respondents hesitating about a pregnancy at increasing risk levels. Within each subgroup a clear relation is found between risk level and the proportion of respondents for whom this risk level would have an influence. But marked differences between the subgroups in their reaction to a

TABLE III. Reactions in the Hypothetical Situation That Both Partners Would Have Been Informed That They Were Carriers of the CF Gene

	Whole group (N = 131)	NC × NC (N = 69)	C × NC (N = 35)	C × ? (N = 25)
If known before marriage				
Decision about children				
- No own children	32%	43%	31%	12%
- Artificial insemination	6%	7%	9%	0%
- Prenatal diagnosis	42%	33%	43%	60%
- Taking the risk	20%	17%	17%	28%
If known while pregnant				
Would use prenatal diagnosis	84%	85%	84%	80%
Would abort CF foetus	56%	62%	49%	48%
Abortion acceptable	78%	85%	71%	64%

given risk level are also observed. The reactions of the 'C × NC' group remarkably resemble those of the 'NC × NC' group at the high risk level, and become more in accordance with those of the 'C × ?' group at the lower risk levels; moreover, the shift takes place at their own objective risk level.

The Jonckheere test for ordered alternatives revealed a significant trend to accept higher risk levels when the objective post test risk was higher ($J^* = 1.91, P < .05$). Multiple comparisons showed that the 'NC × NC' group was less prepared to take the risk than the 'C × ?' group. The curve of the 'C × NC' group did not differ significantly from that of both other groups.

DISCUSSION

Recently, a few studies have addressed the utilization of CF carrier testing by the high-risk population with a family history of CF. Surh et al. [1994] pointed to the overall low participation (less than 10%) in carrier testing by relatives of patients with CF. As possible explanations they suggest both limited dissemination of information through the family and lack of self-perceived risk for being a carrier. Fanos et al. [1995a] identified both structural and psychological barriers to the transmission of genetic information within CF families.

In the line of previous findings [Denayer et al., 1992] knowledge about the recessive transmission of CF was disappointing. A more important question in the context of the present paper however is the knowledge about post test risks. Although 95% of the respondents correctly remembered whether they had a positive or negative test result, only one fourth could situate his post test risk of having a CF child in the correct answer category. Overall, subjects tended to underestimate their risk rather than overestimating it.

Nevertheless, on a group level, the subjects made the distinction between the residual risk in a situation where both partners tested negatively and the residual risk in a situation where one partner is a carrier of the CF gene, and the other one has a negative test result: there was a significant difference between the 'NC × NC' group and the 'C × NC' group for both the mean quantitative estimation and the mean qualitative evaluation of the risk.

The finding that the means in a 'C × ?' situation were not significantly different from the means in a 'C × NC' situation, might lead to the conclusion that these situations are perceived as similar. However, the quantitative estimation as well as the qualitative evaluation of the post test risk showed a high variability within the 'C × ?' group, indicating a very poor understanding of the post test risk of having a CF child in carriers who were tested on an individual basis.

The percentage of subjects unaware of the residual risk was high in all groups. However, not being aware of the limitations of DNA testing especially has implications for carriers whose partner tested negatively: this applies to 36% of the subjects in this subgroup. Fanos et al. [1995b] found that 33% of the interviewed sibs knew that having a CF child was still possible 'if one parent is positive for ΔF508 and the other is negative' while 27% thought that this was not possible. This issue received ample attention in the context of population screening. Watson et al. [1992], who screened 3000 individuals and identified 100 carriers, found that 46% of the carriers whose partner was tested (and had a negative test result) correctly realized there was still a low risk of CF occurring in a pregnancy, while 31% thought there was no risk. These figures are very similar to ours. Miedzybrodzka et al. [1995] on the other hand reported that 79% of the subjects with a negative test result correctly perceived some residual risk of being carriers.

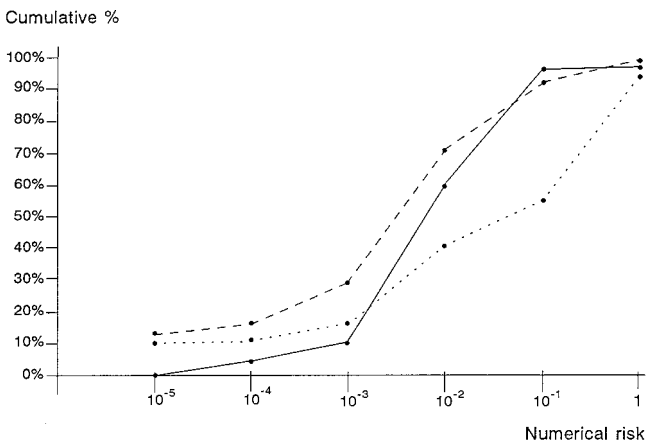

Fig. 1. Proportion of subjects reporting that a specific hypothetical risk of giving birth to a CF child would have an influence. – – – –NC × NC (N = 69);——C × NC (N = 35); ····· C × ? (N = 25).

The fact that a substantial proportion of the 'C × NC' and the 'NC × NC' group was not aware of the limitations of carrier testing is in line with Bekker et al. [1994] who concluded that 'in the longer term, the greater problem would be one of false reassurance'. The psychological literature repeatedly pointed to the difficulties people have in distinguishing small probabilities and to the fact that people tend to categorize a situation as either 'safe' or 'dangerous' [Redelmeier et al., 1993].

Moreover, being aware of the residual risk seems to relate to personality characteristics: the respondents with high scores on trait anxiety were more often aware of the residual risk and perceived this risk as being higher. This correlation with trait anxiety is in line with findings from social psychology: the higher the subjects' trait anxiety, the higher they rate the likelihood that negative events might happen to themselves [Nesse & Klaas, 1994], counteracting the general trend that people perceive their own risk as smaller than the risk of other people [Weinstein, 1989; Welkenhuysen et al., 1996].

The current routine of offering CF carrier testing to CF families clearly underestimates the problems people have in understanding and remembering information about the recessive transmission of CF and the associated risks, and about the limitations of DNA testing. We have no data about the information provided to the subjects when CF carrier testing was offered to them, but the doctors who mediated for carrier testing seem to have relied too much on the process of information transfer within the CF families. CF relatives would obviously take advantage of the educational materials developed in view of the activation of population screening.

We found no difference in reproductive decision making between the group of couples where both partners tested negatively and the group of couples where one partner tested positively while the other one tested negatively. Concerns about the potential anxiety caused in couples in which one partner is a carrier, while the other has a negative test result but can not be excluded as a carrier of a rare CF mutation, seem to be unfounded. On the other hand, we have indications that reproductive decision making was burdensome for those carriers who made childbearing decisions before carrier testing was possible for their partner. Some of them did in fact limit their number of children. The finding that some of the tested subjects were not aware of the availability of carrier testing for their partner shows that an extra effort should be made in this context. The problems encountered in this subgroup draw the attention to the disadvantages of carrier testing in individual subjects. Nevertheless, the majority of the carriers who still were single intended to have their partner tested before starting a pregnancy.

Hypothetical reproductive decisions if the respondents had known before marriage that both carried the CF gene might give some indication about the reaction of carrier couples identified before the occurrence of a CF child. No one of our subjects would consider this an obstacle to marriage. The majority would try to prevent the birth of an affected child, mainly by not having children or by prenatal diagnosis. Although a large majority of the subjects would opt for prenatal diagnosis, and finds selective abortion of a CF foetus acceptable, the proportion who would actually abort a CF foetus was considerably smaller.

It is very interesting to find significant differences among the three subgroups on behalf of this topic: subjects of the 'C × ?' group, for whom this situation is still a possibility, opted significantly more often for having children, with or without prenatal diagnosis, while both other groups, who will never get in this situation, considered more easily remaining childless. The three subgroups also reacted in a different way when asked which risk level would make them hesitate about a pregnancy: there was a significant trend to accept higher risk levels as their own objective risk was higher. Especially the 'NC × NC' group, whose subjects have a minimal post test risk themselves, was less prepared to take a risk than the other groups. These findings suggest that the bare fact of being informed about a 1/4 risk of having a CF child might influence one's attitudes toward reproduction in such a situation. One might expect that carrier couples detected by population screening would primarily opt for prenatal diagnosis in order to avoid the birth of a CF child. However, the actual reproductive decisions of this group remain to be assessed, as well as the reactions of 'carrier × negative test result' couples detected by population screening. The results of this study do suggest that this group will not present major problems.

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APPENDIX

Formulation of the Questionnaire Items Discussed in This Study

What is the risk of a random person to be a carrier of the CF gene?*
1. More than 1 in 10 (more than 10%)
2. Between 1 in 10 and 1 in 100 (between 10% and 1%)
3. Between 1 in 100 and 1 in 1,000 (between 1% and 1 per thousand)
4. Less than 1 in 1,000
5. No risk at all
6. I don't know

The exact risk is ..... (Fill in)

What is the risk of having a CF child when both parents are carriers of the CF gene?*

See first question

What is the risk of having a CF child when one parent is a carrier of the CF gene and the other parent is not?*

See first question

What is the risk of having a CF child when none of the parents carries the CF gene?*

See first question

Who informed you about these risks and about what it means to be a carrier of the CF gene?
1. A family member who? .....
2. My family doctor
3. A gynecologist
4. A pediatrician
5. A doctor in a genetic center
6. Another doctor: .....
7. Newspapers, radio, television

Did your doctor inform you about your residual risk of having a CF child, taking into account the result of your partner?
1. Yes
2. No
3. I don't remember

What is your residual risk of having a CF child, taking into account the result of your partner?*

See first question

How do you evaluate this risk?*
1. A very high risk
2. A high risk
3. A low risk
4. A very low risk
5. No risk at all

Suppose that you were told before marriage that you and your current partner had a 1 in 4 risk of having a CF child in every pregnancy

Would that be a reason to separate?
1. It surely would
2. It probably would
3. It probably would not
4. It surely would not

Suppose that you were told before having children that you and your current partner had a 1 in 4 risk of having a CF child in every pregnancy

What would have been your decision about having children?
1. Remaining childless
2. Using artificial insemination to avoid the birth of a CF child
3. Having own children, and use prenatal diagnosis to avoid the birth of a CF child
4. Having own children, and take the risk

The previous question was about PLANNING a pregnancy. Let's presume now that you were informed about the 1 in 4 risk during the first weeks of a pregnancy

Would you ask for a prenatal diagnosis, in order to know if the child had CF?
1. I surely would
2. I probably would
3. I probably would not
4. I surely would not

Would you ask for an abortion if the result was unfavorable (if the foetus had CF)?

See above

Do you think abortion is acceptable in the case of CF? In other words: is it acceptable for you that other people would have an abortion under such circumstances?
1. Entirely acceptable
2. Rather acceptable
3. Rather not acceptable
4. Not acceptable at all

*For the sake of simplicity we transformed the scale during the analyses so that the highest (subjective) risk received the highest scale value.